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Maryland Department of Health and Mental Hygiene  
Office for Genetics and People with Special Health Care Needs

**Down syndrome (Trisomy 21) Fact Sheet**

**What Is Down syndrome?**

- \* A genetic disorder in which an infant has 47 chromosomes instead of the usual 46

**Signs and Symptoms**

- \* Eyes that slant downward
- \* Small ears that may fold over a little at the top
- \* A small mouth, making the tongue appear to be large
- \* A short neck
- \* Small hands and feet
- \* Weak muscle tone
- \* Short stature in childhood and adulthood
- \*\* Infants born with Down syndrome often have intellectual disability. The degree of the disability varies.

**What is the cause?**

- \* Down syndrome is caused by an extra copy of chromosome 21. It is also called Trisomy 21 because there are three copies of this chromosome instead of two (chromosomes are the structure in cells that contains the genes).

**Is there any treatment?**

- \* Currently, there is no cure for Down syndrome.
- \* Children with Down syndrome need to be followed by their pediatrician regularly since there are several medical concerns that they may have. These include: heart problems, abnormal thyroid function, neck instability, ear infections and hearing problems.
- \* Children with Down syndrome benefit from early intervention services to help with motor and learning skills.

**Living with Down syndrome**

Down syndrome is a lifelong condition. Services early in life will often help babies and children with Down syndrome to improve their physical and intellectual abilities. Most of these services focus on helping children with Down syndrome develop to their full potential. These services include speech, occupational, and physical therapy, and they are typically offered through early intervention programs in each state. Children with Down syndrome may also need extra help or attention in school, although many children are included in regular classes.

Many people with Down syndrome lead productive and fulfilling lives well into adulthood. They can have jobs and live independently. However, it is important for people with Down syndrome to be involved in their community, take good care of themselves, and see a healthcare provider regularly. Families of people with Down syndrome often can help their loved ones by connecting with other families that have had children with Down syndrome. This may help families gain insight into overcoming potential differences in caring for their child with Down syndrome.